

Prenatal diagnosis of a giant intracranial teratoma associated with pulmonary hypoplasia

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Abstract

We present a case in which an intracranial tumour replacing all intracranial contents was diagnosed by sonography at 31 weeks' gestation. The patient was delivered by caesarean section and died shortly after delivery. At necropsy, the tumour was found to be a teratoma with no recognisable normal brain tissue present. Additional findings at necropsy included pulmonary and adrenal hypoplasia. The diagnosis and prognosis of intracranial teratomas diagnosed in utero, and the association of this tumour with pulmonary hypoplasia, are discussed.

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Teratomas are the most common intracranial neoplasms occurring in utero, but are very rare; only about 20 cases have been diagnosed in utero.¹⁻⁷ In most of these cases, sonographic evaluation was prompted in the late second trimester or third trimester by sudden acceleration in the growth of the uterus in a previously uncomplicated pregnancy.^{4,5} Sonographic findings range from hydrocephalus in association with an intracranial mass to macrocephaly with apparent replacement of normal intracranial structures with disorganised solid and cystic tumour masses; extracranial extension has also been reported.⁷ With rare exceptions, intracranial teratomas diagnosed in utero have fatal outcomes. Here, we present a case in which a massive intracranial tumour, presumed to be a teratoma, was diagnosed in utero. The fetus was found at delivery to have pulmonary hypoplasia. We speculate that destruction of midbrain centres controlling res-

piration occurred before 22 weeks' gestation and caused pulmonary hypoplasia.

Case report

A 28 year old G3, P1, SAb1 woman presented at 31 weeks' gestation with a history of accelerated weight gain, rapid growth of the uterine fundus, and dyspnoea. The pregnancy had been previously uncomplicated. A routine ultrasound examination had been performed at 16 weeks' gestation and was reportedly normal, although films were not available for review. There had been no problems noted when the patient was seen at 28 weeks' gestation, except for a 2 cm discrepancy between fundal height and the gestational age. During the next three weeks, the patient experienced a 4.9 kg weight gain with an associated 8 cm increase in fundal height.

At 31 weeks, cursory sonography for evaluation of the size/dates discrepancy suggested hydrocephalus and polyhydramnios. The patient was referred to the Sharp/Children's prenatal diagnostic centre for a detailed sonographic evaluation. Ultrasound showed polyhydramnios, placentomegaly, and macrocephaly. The amniotic fluid index was approximately 40 cm (normal 6 to 24 cm). The placental thickness ranged up to 8 cm, and the subjective impression was that the placental mass was more than twice the normal size for a term pregnancy.

Examination of the head and cranial contents was remarkable. The biparietal diameter was 14.0 cm (normal 7.4 to 7.6 cm). Although the facial bony features and orbits were well preserved, no other normal intracranial structures could be identified; specifically, normal midbrain, cerebellum, ventricles, and cortex were replaced by tumour. The lower two thirds of the cranial cavity was filled with a 'bubbly' appearing solid mass; in the upper third two large cystic structures were present, one on each side of the midline, when the head was viewed in coronal section (fig 1). There were no large areas of calcification, but a variegated pattern of echodensities within the 'bubbly' component was felt to be consistent with numerous small calcifications.

Anatomical visualisation was limited owing to the fetal position, but normal anatomy of the spine, heart, kidneys, liver, and extremities was seen. The stomach bubble was not seen despite favourable orientation of the fetus during the scan. The bladder and genitalia could not be seen because of breech presentation. No fetal movement was present.

The diagnosis of an intracranial tumour, probably a teratoma, was made. Because of

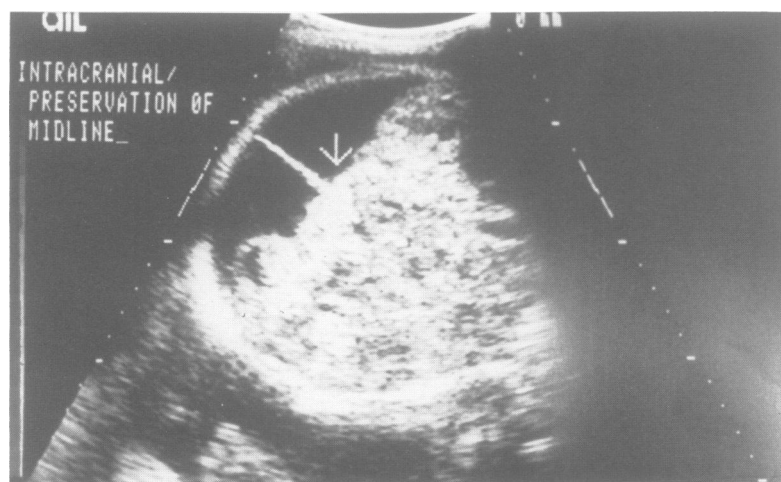


Figure 1 Ultrasound at 31 weeks' gestation. In this coronal view, the cystic component of the tumour filling the upper portion of the cranial vault, and the solid component below, are clearly seen.

increasing maternal discomfort, delivery was planned within the next week. Owing to the size and predominantly solid nature of the mass, as well as the breech presentation, delivery was by classical caesarean section (fig 2). The infant was bradycardic at delivery, had no spontaneous respirations, and was pronounced dead 25 minutes later.

At necropsy, the cranium was massively enlarged and completely filled with a soft, haemorrhagic appearing mass. No normal brain architecture was identified. All three germ layers were represented within the mass, confirming the diagnosis of intracranial teratoma (fig 3). There was no extracranial extension of the tumour.



Figure 2 The infant at birth.

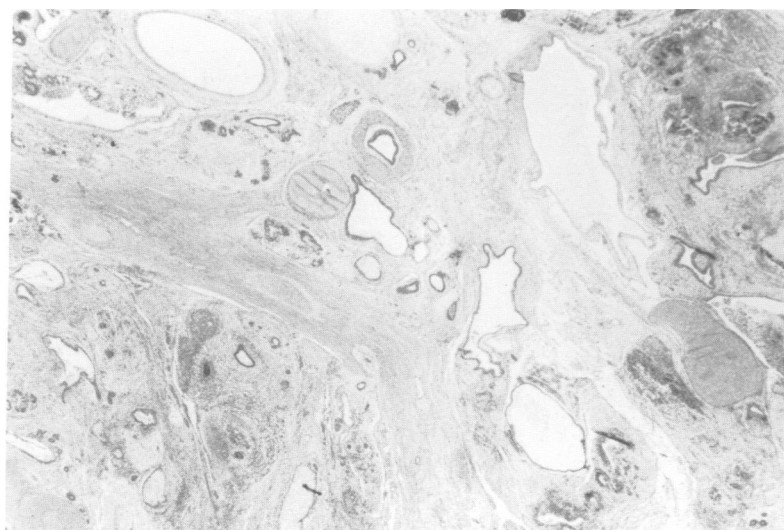


Figure 3 Photomicrograph of a section of the teratoma showing cartilage, neural tissue, smooth muscle, mixed mucinous and serous epithelium, respiratory epithelium, and squamous epithelium.

A gestational age of 32.5 weeks was determined by foot length measurement (6.3 cm). In addition to the intracranial pathology, a diffusely enlarged liver (178 g, normal 76 g), small adrenals (1.6 g, normal 5.0 g), and hypoplastic lungs (24 g, normal 38 ± 10 g) were noted. Microscopic examination showed reduced radial alveolar counts, confirming the diagnosis of pulmonary hypoplasia.

The placenta was large (570 g, normal 290 to 320 g) and spongy. Microscopic examination showed diffuse chorionic villus oedema and pigmented macrophages within the amnion. Chorionic villus oedema has been reported in association with sacrococcygeal teratomas, but not with intracranial teratomas. The fetus and the teratoma both had 46,XX chromosome complements, with identical pericentromeric markers.

The mother has subsequently had a term delivery of a normal infant.

Discussion

Teratomas are the most common fetal tumours, with intracranial teratomas second in frequency only to sacrococcygeal teratomas.⁸⁻¹⁰ Teratomas represent half of all neonatal intracranial neoplasms.⁴

There are 20 previous reports of prenatal diagnosis of intracranial teratomas¹⁻⁷ and among these there was only one survivor. This infant had complete resection of a malignant intracranial teratoma, but has severe intellectual impairment.⁵ The sonographic diagnosis of intracranial teratoma has usually occurred in the setting of rapid increase in the fundal height during the late second and third trimester; sonography shows polyhydramnios and either macrocephaly with a large intracranial tumour, or hydrocephalus and a mass lesion. In cases with diagnosis several weeks before delivery, increase in the size of the mass and of the fetal head has been dramatic.²

The differential diagnosis of fetal intracranial masses includes teratoma, neuroectodermal tumours, astrocytomas, glioblastoma multiform, and choroid plexus papilloma.⁶ Additionally, there has been one report in which intracranial teratoma was suspected, but the fetus proved to have evolving hydranencephaly.¹¹ There have only been a handful of sonographic diagnoses of intracranial tumours other than teratomas in utero,¹ and differentiation of teratoma from other tumours cannot currently be made with certainty until after delivery.

Extracranial abnormalities noted at necropsy in cases of congenital intracranial teratomas have not been well described. In the present case, extracranial features included hepatomegaly, adrenal hypoplasia, and pulmonary hypoplasia. Odell *et al*² described 'congestive hepatomegaly' in one case and 'pulmonary immaturity' in the other. The degree of pulmonary hypoplasia seen in this case has not been described before.

Proposed mechanisms of the pathogenesis of pulmonary hypoplasia include extrinsic restraint, through space occupying lesions such

as in diaphragmatic hernia, lack of amniotic fluid as in prolonged oligohydramnios, and lack of fetal breathing movements such as in the fetal akinesia sequence.¹² It is believed that not only the presence of amniotic fluid, but also movement of the fetal diaphragm, is necessary for normal lung growth. In the case presented here, we speculate that damage or destruction of the central control mechanisms for fetal breathing occurred well before the clinical presentation and resulted in lung hypoplasia through lack of movement of the diaphragm. The adrenal hypoplasia would also be consistent with early destruction of pituitary tissue as this is a common feature in anencephalic infants.

Intracranial teratomas diagnosed in utero have an almost uniformly fatal outcome. This case shows the rapidly destructive nature of these tumours as the pituitary and brainstem functions appear to have been affected before clinical symptoms appeared.

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